SINGLE NUCLEOTIDE... Title:

Inventors: Eric S. Lander, et al.

HT1220 Report

RECORD INFORMATION

1220 Gene ID: 1220 Sequence ID: 1220 Protein ID:

thrombospondin 1, alt. transcript 1 Sequence name:

Genome: nucleus Homo sapiens Taxon: 1220

Locus:

thrombospondin 1 Common Name:

40 Role ID:

Coding sequence length: 3513 nt Transcript sequence length: 5722 nt THC201673 Expression data:

ACCESSION DATA

HT1220 is derived from accessions(s):

```
SP: P07996 (THROMBOSPONDIN 1 PRECURSOR.)
GB:X04665 (Human mRNA for thrombospondin)
GB:X14787 (Human mRNA for thrombospondin)
GB:U12471(thrombospondin-p50 {Homo sapiens})
GB:M99425 (Human thrombospondin mRNA, 3' end.)
PIR:G01478 (thrombospondin-p50 - human (fragment))
GB: U12471 (Human thrombospondin-1 gene, partial cds.)
GB:J04835 (Human thrombospondin gene, exons 1, 2 and 3.)
GB:M25631 (Homo sapiens (clone lambda-TS-33) thrombospondin (THBS) mRNA, 5' end.)
```

ALTERNATIVE SPLICE INFORMATION

Alternative splice forms for this gene:

HT3987 thrombospondin 1, alt. transcript 2

MAPPING DATA

GDB accession(s) for this gene:

Symbol GDB ID:

Figure 1A

Title: SINGLE NUCLEOTIDE...

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gdb:120438 THBS1

cDNA FEATURES

Feature	End 5	End 3
coding_seq 3'UT spjunc_h	112 3625 1235	3624 5722 1236

SEQUENCE

nucleotide:

ggacgcacaggcattccccgcgcccctccagccctcgccgccctcgccaccgctcccggc cgccgcgctccggtacacacaggatccctgctgggcaccaacagctccaccatggggctg tetggeggagacaacagegtgtttgacatetttgaacteaceggggeegeeegeaagggg tetgggegeegaetggtgaagggeeeegaeeetteeageeeagettteegeategaggat gccaacctgatcccccctgtgcctgatgacaagttccaagacctggtggatgctgtgcgg gcagaaaagggtttcctccttctggcatccctgaggcagatgaagaagacccggggcacg ctgctggccctggagcggaaagaccactctggccaggtcttcagcgtggtgtccaatggc aaggegggcaccetggacetcageetgacegtecaaggaaageageaegtggtgtetgtg gaagaagctctcctggcaaccggccagtggaagagcatcaccctgtttgtgcaggaagac agggcccagctgtacatcgactgtgaaaagatggagaatgctgagttggacgtccccatc caaagcgtcttcaccagagacctggccagcatcgccagactccgcatcgcaaagggggc gtcaatgacaatttccagggggtgctgcagaatgtgaggtttgtctttggaaccacacca gaagacateeteaggaacaaaggetgeteeagetetaeeagtgteeteeteaceettgae aaggacttgcaagccatctgcggcatctcctgtgatgagctgtccagcatggtcctggaa ctcaggggcctgcgcaccattgtgaccacgctgcaggacagcatccgcaaagtgactgaa gagaacaaagagttggccaatgagctgaggcggcctcccctatgctatcacaacggagtt cagtacagaaataacgaggaatggactgttgatagctgcactgagtgtcactgtcagaac tcagttaccatctgcaaaaaggtgtcctgcccatcatgccctgctccaatgccacagtt cctgatggagaatgctgtcctcgctgttggcccagcgactctgcggacgatggctggtct ccatggtccgagtggacctcctgttctacgagctgtggcaatggaattcagcagcggc cgctcctgcgatagcctcaacaaccgatgtgagggctcctcggtccagacacggacctgc cacattcaggagtgtgacaaaagatttaaacaggatggtggctggagccactggtccccg tggtcatcttgttctgtgacatgtggtgatggtgatcacaaggatccggctctgcaac tctcccagcccccagatgaatgggaaaccctgtgaaggcgaagcgcgggagaccaaagcc tgcaagaaagacgcctgccccatcaatggaggctggggtccttggtcaccatgggacatc tgttctgtcacctgtggaggagggtacagaaacgtagtcgtctctgcaacaaccccgca ccccagtttggaggcaaggactgcgttggtgatgtaacagaaaccagatctgcaacaag caggactgtccaattgatggatgcctgtccaatccctgctttgccggcgtgaagtgtact agctaccctgatggcagctggaaatgtggtgcttgtccccctggttacagtggaaatggc atccagtgcacagatgttgatgagtgcaaagaagtgcctgatgcctgcttcaaccacaat ttcaccggctcacagcccttcggccagggtgtcgaacatgccacggccaacaacaggtg tgcaagccccgtaacccctgcacggatgggacccacgactgcaacaagaacgccaagtgc aactacctgggccactatagcgaccccatgtaccgctgcgagtgcaagcctggctacgct gtgtgcgtggccaatgcgacttaccactgcaaaaaggataattgccccaaccttcccaac tcagggcaggaagactatgacaaggatggaattggtgatgcctgtgatgatgacgatgac aatgataaaattccagatgacagggacaactgtccattccattacaacccagctcagtat gactatgacagagatgatgtgggagaccgctgtgacaactgtccctacaaccacaaccca

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gatcaggcagacacagacaacaatggggaaggaggcctgtgctgcagacattgatgga gacggtatcctcaatgaacgggacaactgccagtacgtctacaatgtggaccagagagac actgatatggatggggttggagatcagtgtgacaattgccccttggaacacaatccggat cagotggáototgaotoagaoogoattggagatacotgtgacaacaatcaggatattgat gaagatggccaccagaacaatctggacaactgtccctatgtgcccaatgccaaccaggct gaccatgacaaagatggcaagggagatgcctgtgaccacgatgatgacaacgatggcatt cctgatgacaaggacaactgcagactcgtgcccaatcccgaccagaaggactctgacggc gatggtcgaggtgatgcctgcaaagatgattttgaccatgacagtgtgccagacatcgat gacatetgteetgagaatgttgacateagtgagaeegattteegeegatteeagatgatt cctctggaccccaaagggacatcccaaaatgaccctaactgggttgtacgccatcagggt aaagaactcgtccagactgtcaactgtgatcctggactcgctgtaggttatgatgagttt aatgetgtggactteagtggcacettetteatcaacacegaaagggacgatgactatget ggatttgtctttggctaccagtccagcagccgcttttatgttgtgatgtggaagcaagtc acccagtectactgggacaccaaccccacgagggeteagggatactegggeetttötgtg aaagttgtaaactccaccacagggcctggcgagcacctgcggaacgccctgtggcacaca ggaaacacccctggccaggtgcgcaccctgtggcatgaccctcgtcacataggctggaaa gatttcaccgcctacagatggcgtctcagccacaggccaaagacgggtttcattagagtg gtgatgtatgaagggaagaaatcatggctgactcaggacccatctatgataaaacctat getggtggtagaetagggttgtttgtetteteteagaaatggtgttettetetgaeetg aatgctggtattgcaccttctggaactatgggcttgagaaaacccccaggatcacttctc cttggcttccttctttctgtgcttgcatcagtgtggactcctagaacgtgcgacctgcc tcaagaaaatgcagttttcaaaaacagactcatcagcattcagcctccaatgaataagac atcttccaagcatataaacaattgctttggtttccttttgaaaaagcatctacttgcttc agttgggaaggtgcccattccactctgcctttgtcacagagcagggtgctattgtgaggc catctctgagcagtggactcaaaagcattttcaggcatgtcagagaagggaggactcact agaattagcaaacaaaaccaccctgacatcctccttcaggaacacggggagcagaggcca aagcactaaggggagggcgcatacccgagacgattgtatgaagaaaatatggaggaactg ttacatgttcggtactaagtcattttcaggggattgaaagactattgctggatttcatga tgctgactggcgttagctgattaacccatgtaaataggcacttaaatagaagcaggaaag ggagacaaagactggcttctggacttcctccctgatccccacccttactcatcaccttgc ctggtcacattgaaattggtggcttcattctagatgtagcttgtgcagatgtagcaggaa aataggaaaacctaccatctcagtgagcaccagctgcctcccaaaggaggggcagccgtg ttctcttttttccgtaattactaggtagttttctaattctctcttttggaagtatgattt ttttaaagtctttacgatgtaaaatatttattttttacttattctggaagatctggctga aggattattcatggaacaggaagaagcgtaaagactatccatgtcatctttgttgagagt cttcgtgactgtaagattgtaaatacagattatttattaactctgttctgcctggaaatt taggetteataeggaaagtgtttgagageaagtagttgaeatttateageaaatetettg caagaacagcacaaggaaaatcagtctaataagctgctctgccccttgtgctcagagtgg atgttatgggattccttttttctctgttttatcttttcaagtggaattagttggttatcc atttgcaaatgttttaaattgcaaagaaagccatgaggtcttcaatactgttttacccca aaaagagaaaaaaatgacaaaaggtgaaacttacatacaaatattacctcatttgttgtg tgactgagtaaagaatttttggatcaagcggaaagagtttaagtgtctaacaaacttaaa gctactgtagtacctaaaaagtcagtgttgtacatagcataaaaactctgcagagaagta ttcccaataaggaaatagcattgaaatgttaaatacaatttctgaaagttatgtttttt totatoatotggtatacoattgotttatttttataaattattttctoattgcoattggaa tagaatattcagattgtgtagatatgctatttaaataatttatcaggaaatactgcctgt agagttagtatttctattttatataatgtttgcacactgaattgaagaattgttggttt tacattctaaagcagtgtaagttgtatattactgtttcttatgtacaaggaacaacaata aatcatatggaaatttatattt

protein:

 ${\tt MGLAWGLGVLFLMHVCGTNRIPESGGDNSVFDIFELTGAARKGSGRRLVKGPDPSSPAFR}$

Title: SINGLE NUCLEOTIDE...

Inventors: Eric S. Lander, et al.

IEDANLIPPVPDDKFQDLVDAVRAEKGFLLLASLRQMKKTRGTLLALERKDHSGQVFSVV SNGKAGTLDLSLTVQGKQHVVSVEEALLATGQWKSITLFVQEDRAQLYIDCEKMENAELD VPIQSVFTRDLASIARLRIAKGGVNDNFQGVLQNVRFVFGTTPEDILRNKGCSSSTSVLL TLDNNVVNGSSPAIRTNYIGHKTKDLQAICGISCDELSSMVLELRGLRTIVTTLQDSIRK VTEENKELANELRRPPLCYHNGVQYRNNEEWTVDSCTECHCQNSVTICKKVSCPIMPCSN ATVPDGECCPRCWPSDSADDGWSPWSEWTSCSTSCGNGIQQRGRSCDSLNNRCEGSSVQT RTCHIQECDKRFKQDGGWSHWSPWSSCSVTCGDGVITRIRLCNSPSPQMNGKPCEGEARE TKACKKDACPINGGWGPWSPWDICSVTCGGGVQKRSRLCNNPAPQFGGKDCVGDVTENQI CNKQDCPIDGCLSNPCFAGVKCTSYPDGSWKCGACPPGYSGNGIQCTDVDECKEVPDACF NHNGEHRCENTDPGYNCLPCPPRFTGSQPFGQGVEHATANKQVCKPRNPCTDGTHDCNKN AKCNYLGHYSDPMYRCECKPGYAGNGIICGEDTDLDGWPNENLVCVANATYHCKKDNCPN LPNSGQEDYDKDGIGDACDDDDDDKIPDDRDNCPFHYNPAQYDYDRDDVGDRCDNCPYN ${\tt HNPDQADTDNNGEGDACAADIDGDGILNERDNCQYVYNVDQRDTDMDGVGDQCDNCPLEH}$ ${\tt NPDQLDSDSDRIGDTCDNNQDIDEDGHQNNLDNCPYVPNANQADHDKDGKGDACDHDDDN}$ DGIPDDKDNCRLVPNPDQKDSDGDGRGDACKDDFDHDSVPDIDDICPENVDISETDFRRF QMIPLDPKGTSQNDPNWVVRHQGKELVQTVNCDPGLAVGYDEFNAVDFSGTFFINTERDD DYAGFVFGYQSSSRFYVVMWKQVTQSYWDTNPTRAQGYSGLSVKVVNSTTGPGEHLRNAL WHTGNTPGQVRTLWHDPRHIGWKDFTAYRWRLSHRPKTGFIRVVMYEGKKIMADSGPIYD KTYAGGRLGLFVFSQEMVFFSDLKYECRDP

Title: SINGLE NUCLEOTIDE...

Inventors: Eric S. Lander, et al.

HT2143 Report

RECORD INFORMATION

Gene ID: 2081
Sequence ID: 2143
Protein ID: 2125

Sequence name: thrombospondin 4

Genome: nucleus
Taxon: Homo sapiens

Locus: 2081

Common Name: thrombospondin 4

Role ID: 4

Coding sequence length: Transcript sequence length: Expression data: 2886 nt 3074 nt THC168897

ACCESSION DATA

HT2143 is derived from accessions(s):

SP:P35443 (THROMBOSPONDIN 4 PRECURSOR.)
GB:Z19585(thrombospondin-4 {Homo sapiens})
GB:Z19585(H.sapiens mRNA for thrombospondin-4)
PIR:A55710 (thrombospondin 4 precursor - human)

cDNA FEATURES

Feature		End	5	End	3
				- -	
coding_seq		28		2913	3
3'UT	~	2914		3074	Į

SEQUENCE

nucleotide:

gaattooggggagcaggaagagccaacatgctggccccgcgggagccgccgtcctcctg ctgcacctggtcctgcagcggtggctagcggcaggcgccaggccaccccccaggtcttt gaccttctcccatcttccagtcagaggctaaacccaggcgtcttgctgccagtcctgaca gaccccgcctgaatgatctctatgtgatttccaccttcaagctgcagactaaaagttca gccaccatcttcggtctttactcttcaactgacaacagtaaatattttgaatttactgtgatgggacgcttaagcaaagccatcttc

Title: SINGLE NUCLEOTIDE...

Inventors: Eric S. Lander, et al.

gtggttttcaacaacctgcagctggcagacggaaggcggcacaggatcctcctgaggctg agcaatttgcagcgaggggccggctccctagagctctacctggactgcatccaggtggat tccgttcacaatctccccagggcctttgctggcccctcccagaaacctgagaccattgaa ttgaggactttccagaggaagccacaggacttcttggaagagctgaagctggtggtgaga ggctcactgttccaggtggccagcctgcaagactgcttcctgcagcagagtgagccactg gctgccacaggcacaggggactttaaccggcagttcttgggtcaaatgacacaattaaac caactcctgggagaggtgaaggaccttctgagacagcaggttaaggaaacatcatttttg cgaaacaccatagctgaatgccaggcttgcggtcctctcaagtttcagtctccgacccca agcacggtggtcgccccggctccccctgcaccgccaacacgcccacctcgtcggtgtgac tccaacccatgtttccgaggtgtccaatgtaccgacagtagagatggcttccagtgtggg ccctgcccgagggctacacaggaaacgggatcacctgtattgatgttgatgagtgcaaa taccatccctgctacccgggcgtgcactgcataaatttgtctcctggcttcagatgtgac gcctgcccagtgggcttcacagggcccatggtgcagggtgttgggatcagttttgccaag tcgatctgcgttaatactttgggatcttaccgctgtgggccttgtaagccggggtatact ggtgatcagataaggggatgcaaagtggaaagaaactgcagaaacccagagctgaaccct qtcqqttqqqctqqaqatqqctatatctqtqqaaaqqatqtqqacatcqacaqttacccc gacgaagaactgccatgctctgccaggaactgtaaaaaaggacaactgcaaatatgtgcca aattctggccaagaagatgcagacagagatggcattggcgacgcttgtgacgaggatgct gacggagatgggatcctgaatgagcaggataactgtgtcctgattcataatgtggaccaa aggaacagcgataaagatatctttggggatgcctgtgataactgcctgagtgtcttaaat aacgaccagaaagacaccgatggggatggaagaggagatgcctgtgatgatgacatggat ggagatggaataaaaaacattctggacaactgcccaaaatttccccaatcgtgaccaacgg qacaaqqatggtgatggtgtgggggatgcctgtgacagttgtcctgatgtcagcaaccct aaccagtctgatgtggataatgatctggttggggactcctgtgacaccaatcaggacagt gatggagatgggcaccaggacagcacagacaactgccccaccgtcattaacagtgcccag ctggacaccgataaggatggaattggtgacgagtgtgatgatgatgatgacaatgatggt atcccagacctggtgccccctggaccagacaactgccggctggtccccaacccagcccag gaggatagcaacagcgacggagtgggagacatctgtgagtctgactttgaccaggaccag gtcatcgatcgatcgacgtctgcccagagaacgcagaggtcaccctgaccgacttcagg gtcctgaaccagggcatggagattgtacagaccatgaacagtgatcctggcctggcagtg gggtacacagettttaatggagttgacttcgaagggacettccatgtgaatacecagaca gatgatgactatgcaggetttatettttggetaccaagatagetecagettetacgtggte atgtggaagcagacggagcagacatattggcaagccaccccattccgagcagttgcagaa cctggcattcagctcaaggctgtgaagtctaagacaggtccaggggagcatctccggaac tccctqtqqcacacqqqqqacaccaqtqaccaqqtcaqqctgctqtqqaaqqactccaqq aatgtgggctggaaggacaaggtgtcctaccgctggttcctacagcacaggccccaggtg ggctacatcagggtacgattttatgaaggctctgagttggtggctgactctggcgtcacc $\verb|atagacaccacaatigcgtggaggccgacttggcgttttctgcttctctcaagaaaacatc|$ atotggtocaacotcaagtatogotgcaatgacacoatccotgaggacttccaagagttt caaacccagaatttcgaccgcttcgataattaaaccaaggaagcaatctgtaactgcttt tcqqaacactaaaaccatatatattttaacttcaattttctttagcttttaccaacccaa atatatcaaaacgttttatgtgaatgtggcaataaaggagaagagatcatttttaaaaaa aaaaaaaaaaaa

protein:

MLAPRGAAVLLLHLVLQRWLAAGAQATPQVFDLLPSSSQRLNPGALLPVLTDPALNDLYV ISTFKLQTKSSATIFGLYSSTDNSKYFEFTVMGRLSKAILRYLKNDGKVHLVVFNNLQLA DGRRHRILLRLSNLQRGAGSLELYLDCIQVDSVHNLPRAFAGPSQKPETIELRTFQRKPQ DFLEELKLVVRGSLFQVASLQDCFLQQSEPLAATGTGDFNRQFLGQMTQLNQLLGEVKDL LRQQVKETSFLRNTIAECQACGPLKFQSPTPSTVVAPAPPAPPTRPPRRCDSNPCFRGVQ CTDSRDGFQCGPCPEGYTGNGITCIDVDECKYHPCYPGVHCINLSPGFRCDACPVGFTGP MVQGVGISFAKSNKQVCTDIDECRNGACVPNSICVNTLGSYRCGPCKPGYTGDQIRGCKV ERNCRNPELNPCSVNAQCIEERQGDVTCVCGVGWAGDGYICGKDVDIDSYPDEELPCSAR NCKKDNCKYVPNSGQEDADRDGIGDACDEDADGDGILNEQDNCVLIHNVDQRNSDKDIFG DACDNCLSVLNNDQKDTDGDGRGDACDDDMDGDGIKNILDNCPKFPNRDQRDKDGDGVGD

Title: SINGLE NUCLEOTIDE...

Inventors: Eric S. Lander, et al.

ACDSCPDVSNPNQSDVDNDLVGDSCDTNQDSDGDGHQDSTDNCPTVINSAQLDTDKDGIG DECDDDDDDGIPDLVPPGPDNCRLVPNPAQEDSNSDGVGDICESDFDQDQVIDRIDVCP ENAEVTLTDFRAYQTVGLDPEGDAQIDPNWVVLNQGMEIVQTMNSDPGLAVGYTAFNGVD FEGTFHVNTQTDDDYAGFIFGYQDSSSFYVVMWKQTEQTYWQATPFRAVAEPGIQLKAVK SKTGPGEHLRNSLWHTGDTSDQVRLLWKDSRNVGWKDKVSYRWFLQHRPQVGYIRVRFYE GSELVADSGVTIDTTMRGGRLGVFCFSQENIIWSNLKYRCNDTIPEDFQEFQTQNFDRFD N

Figure 2C

App No.: 09/657,472
Title: SINGLE NUCLEOTIDE...
Inventors: Eric S. Lander, *et al*.

	
All AA	o:a.
Rol AA	<u>z 4</u>
ARNI	<u>0,0</u>
Mutation T'Ref NT	A .0
Mutation	Missense A
	CCAACT
	TOBATGGCTGGCCCA AATGAGAACCTGGTGTG M
	CAJA/GICG
Seq	CGAAATB
Flanking Sec	TOGATE
	, Informbospondin 4 TOGATGGCTGGCCCAJAGTIG
Description	thrombos informbos
osklon Gene L	2210 THBS1 1186 THBS4
Posid	NA 1
G Golde	G33414 IITHT1220 mRNA G35512 11THT2143 mRNA
Upes	G334:14 11THT12
Polv	G334114